Statutory Approvals Committee – minutes

Centre 0102 (Guys Hospital)

Pre-implantation Genetic Diagnosis (PGD) application for Scapuloperoneal spinal muscular atrophy (SPSMA), OMIM #181405

Thursday, 17 December 2020

HFEA, 2nd Floor, 2 Redman Place, London E20 1JQ via Teams Meeting

Committee members	Margaret Gilmore (Chair) Emma Cave Anne Lampe Ruth Wilde	
Members of the Executive	Moya Berry Catherine Burwood	Committee Officer Licensing Manager
Specialist Adviser	Dr. Alison Male	
Legal Adviser	Tom Rider	Fieldfisher - LLP

Declarations of interest:

- Members of the committee declared that they had no conflicts of interest in relation to this item.

The committee had before it:

- 9th edition of the HFEA Code of Practice.
- Standard licensing and approvals pack for committee members.

The following papers were considered by the committee:

- Executive summary
- PGD application form
- Redacted peer review
- Genetic Alliance statement
- Public view on the application
- Licence Committee Minutes 26 January 2012 – PGD Application for Charcot Marie Tooth Disease Type 2, OMIM #609260

1. Consideration of application

1.1. The committee welcomed the advice of its specialist adviser, Dr. Alison Male, who confirmed that the condition was as described in the papers.

1.2. The committee noted that the description in the PGD application for Scapuloperoneal spinal muscular atrophy (SPSMA), OMIM #181405, is consistent with the peer review.

1.3. The committee noted that the condition being applied for is not on the list of approved PGD conditions.

1.4. The committee noted that the Genetic Alliance statement provided a perspective on the impact of the condition on patients, their families and carers.

1.5. The committee had regard to its decision tree. The committee noted that the centre is licensed to carry out PGD. The committee was also satisfied that the centre has experience of carrying out PGD and that generic patient information about its PGD programme and associated consent forms had previously been received by the HFEA.

1.6. The committee noted that the proposed purpose of testing the embryos was as set out in paragraph 1ZA(1)(b) of Schedule 2 of the Act, i.e. 'where there is a particular risk that the embryo may have any gene, chromosome or mitochondrion abnormality, establishing whether it has that abnormality or any other gene, chromosome or mitochondrion abnormality'.

1.7. The committee noted that Scapuloperoneal spinal muscular atrophy (SPSMA), OMIM #181405, is inherited in an autosomal dominant manner, which means there is a 50% chance of an embryo being affected by the condition in each pregnancy if either parent has a relevant mutation.

1.8. The committee noted the penetrance for the condition is incomplete, with some individuals only having mild symptoms of muscle weakness, while others may require full-time wheelchair use from childhood, and/or a tracheostomy for breathing assistance.

1.9. Scapuloperoneal spinal muscular atrophy (SPSMA), OMIM #181405, is characterised by progressive muscle weakness, particularly in the muscles in the shoulder blades and lower legs. Affected individuals can develop weakness of the voice box, which can cause issues with speaking or breathing. Some individuals may also have changes to the skeleton, such as curving of the spine and others can be born with missing muscles.

1.10. There is no cure for the condition and symptoms are managed with therapies and surgery.

1.11. The committee noted the executive's request to consider Scapuloperoneal spinal muscular atrophy (SPSMA), OMIM #181405, for inclusion on the list of conditions approved for PGD. The committee agreed to consider the application on this basis.

1.12. The committee also noted the recommendation of the peer reviewer to consider two other types of conditions for inclusion on the list for which PGD can be applied. These are Hereditary motor and sensory neuropathy, type IIc (HMSN2C), OMIM #606071, (also known as Charcot-Marie-Tooth disease, type 2c), and Neuronopathy, distal hereditary motor, type VIII (HMN8) OMIM #600175 (also known as Spinal muscular atrophy, distal, congenital non progressive).

1.13. These conditions are of a similar phenotype to Scapuloperoneal spinal muscular atrophy (SPSMA), OMIM #181405 and are both inherited in an autosomal dominant manner. The conditions can present from birth and are characterised by progressive muscle weakness and atrophy, vocal cord anomalies, impairment of the respiratory muscles, sensorineural hearing loss and distal hands and feet weakness.

2. Decision

2.1. The committee considered that, in the worst-case scenario, Scapuloperoneal spinal muscular atrophy (SPSMA), OMIM #181405, a rare, neuromuscular condition, is potentially life limiting

and can present from birth/early childhood with severe muscle weakness. The condition interferes with movement and may be severe enough to affect daily activities of everyday life. In some cases, those affected may be wheelchair bound and some may require mechanical assistance for breathing. There is no cure for the condition and the committee considered the potentially severe physical impact on those affected with the condition.

2.2. The committee considered the conditions Hereditary motor and sensory neuropathy, type IIc (HMSN2C), OMIM #606071 and Neuronopathy, distal hereditary motor, type VIII (HMN8) OMIM #600175, are of a similar phenotype and presentation to Scapuloperoneal spinal muscular atrophy (SPSMA), OMIM #181405. The conditions can present from birth and in the worst-case scenario severely affect mobility, breathing, hearing, which may severely impact on the quality of life of those affected.

2.3. The committee had regard to its explanatory note and confirmed that, on the basis of the information presented, it was satisfied that there is a particular risk that an embryo may have the abnormality in question and that there is a significant risk that a person with such an abnormality will, given the condition's worst symptoms, have or develop a serious physical disability, a serious illness or any other serious medical condition.

2.4. The committee was therefore satisfied that the following conditions meet the criteria for testing under paragraph 1ZA(1)(b) and (2) of Schedule 2 of the Act.

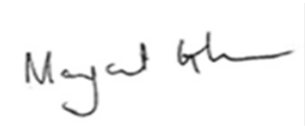
2.5. The committee agreed to authorise testing for:

- Scapuloperoneal spinal muscular atrophy (SPSMA), OMIM #181405
- Hereditary motor and sensory neuropathy, type IIc (HMSN2C), OMIM #606071
- Neuronopathy, distal hereditary motor, type VIII (HMN8), OMIM #600175

3. Chairs signature

3.1. I confirm this is a true and accurate record of the meeting.

Signature



Name
Margaret Gilmore

Date
14 January 2021